Feingold syndrome

Feingold syndrome is a disorder that affects many parts of the body. The signs and symptoms of this condition vary among affected individuals, even among members of the same family.

Individuals with Feingold syndrome have characteristic abnormalities of their fingers and toes. Almost all people with this condition have a specific hand abnormality called brachymesophalangy, which refers to shortening of the second and fifth fingers. Other common abnormalities include fifth fingers that curve inward (clinodactyly), underdeveloped thumbs (thumb hypoplasia), and fusion (syndactyly) of the second and third toes or the fourth and fifth toes.

People with Feingold syndrome are frequently born with a blockage in part of their digestive system called gastrointestinal atresia. In most cases, the blockage occurs in the esophagus (esophageal atresia) or in part of the small intestine (duodenal atresia). Additional common features of Feingold syndrome include an unusually small head size (microcephaly), a small jaw (micrognathia), a narrow opening of the eyelids (short palpebral fissures), and mild to moderate learning disability. Less often, affected individuals have hearing loss, impaired growth, and kidney and heart abnormalities.

Frequency

Feingold syndrome appears to be a rare condition, although its exact prevalence is unknown.

Genetic Changes

Mutations in the *MYCN* gene cause Feingold syndrome. This gene provides instructions for making a protein that plays an important role in the formation of tissues and organs during embryonic development. Studies in animals suggest that this protein is necessary for normal development of the limbs, heart, kidneys, nervous system, digestive system, and lungs. The MYCN protein regulates the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, this protein is called a transcription factor.

Mutations in the *MYCN* gene that cause Feingold syndrome prevent one copy of the gene in each cell from producing any functional MYCN protein. As a result, only half the normal amount of this protein is available to control the activity of specific genes during embryonic development. It remains unclear how a reduced amount of the MYCN protein causes the specific features of Feingold syndrome.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- microcephaly-mesobrachyphalangy-tracheoesophageal fistula (MMT) syndrome
- microcephaly-oculo-digito-esophageal-duodenal (MODED) syndrome
- oculo-digito-esophagoduodental (ODED) syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Feingold syndrome 1
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0796068/
- Genetic Testing Registry: Feingold syndrome 2
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280489/

Other Diagnosis and Management Resources

- GeneReview: Feingold Syndrome 1
  https://www.ncbi.nlm.nih.gov/books/NBK7050
- MedlinePlus Encyclopedia: Duodenal Atresia
  https://medlineplus.gov/ency/article/001131.htm
- MedlinePlus Encyclopedia: Esophageal Atresia
  https://medlineplus.gov/ency/article/000961.htm
- MedlinePlus Encyclopedia: Microcephaly
  https://medlineplus.gov/ency/article/003272.htm
- MedlinePlus Encyclopedia: Webbing of the Fingers or Toes
  https://medlineplus.gov/ency/article/003289.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html

- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Duodenal Atresia
  https://medlineplus.gov/ency/article/001131.htm
- Encyclopedia: Esophageal Atresia
  https://medlineplus.gov/ency/article/000961.htm
- Encyclopedia: Microcephaly
  https://medlineplus.gov/ency/article/003272.htm
- Encyclopedia: Webbing of the Fingers or Toes
  https://medlineplus.gov/ency/article/003289.htm
- Health Topic: Foot Injuries and Disorders
  https://medlineplus.gov/footinjuriesanddisorders.html
- Health Topic: Hand Injuries and Disorders
  https://medlineplus.gov/handinjuriesanddisorders.html
- Health Topic: Intestinal Obstruction
  https://medlineplus.gov/intestinalobstruction.html

Genetic and Rare Diseases Information Center

- Feingold syndrome
  https://rarediseases.info.nih.gov/diseases/8407/feingold-syndrome

Educational Resources

- Boston Children's Hospital: Thumb Hypoplasia/ Aplasia
  http://www.childrenshospital.org/conditions-and-treatments/conditions/thumb-hypoplasia-aplasia
- Disease InfoSearch: FEINGOLD SYNDROME 2
  http://www.diseaseinfosearch.org/FEINGOLD+SYNDROME+2/8426
- MalaCards: feingold syndrome
  http://www.malacards.org/card/feingold_syndrome

- New York-Presbyterian Hospital: Congenital Hand Deformities
  http://www.nyp.org/library/85%257CP01120
- Orphanet: Feingold syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1305

Patient Support and Advocacy Resources

- Resource List from the University of Kansas: Limb Anomalies
  http://www.kumc.edu/gec/support/limb.html

GeneReviews

- Feingold Syndrome 1
  https://www.ncbi.nlm.nih.gov/books/NBK7050

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28feingold+syndrome%5BTIAB%5D%29+OR+%28microcephaly-oculo-digito-esophageal-duodenal+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- FEINGOLD SYNDROME 1
  http://omim.org/entry/164280

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  https://www.ncbi.nlm.nih.gov/books/NBK7050
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